Holocarboxylase synthetase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Holocarboxylase synthetase deficiency. ORPHA:79242

Holocarboxylase synthetase (HCS) deficiency is a life-threatening early-onset form of multiple carboxylase deficiency (see this term), an inborn error of biotin metabolism, that, if untreated, is characterized by vomiting, tachypnea, irritability, lethargy, exfoliative dermatitis, and seizures that can worsen to coma.